SUPPLEMENTARY TEXT

PATHOPHYSIOLOGY

The pathophysiology is thought to be due to a dysfunction in the innate and adaptive immune system resulting in the loss of 'self-tolerance'.[1] Subsequently, this results in B cell activation and increased autoantibody production. Additionally, there is a development of and failure of the removal of immune complexes from the circulation causing deposition in the body organs. Certain medications such as hydralazine, methyldopa, and isoniazid has been reported to induce SLE with positive anti-histone and negative anti-DNA antibodies. Ultraviolet light, viral infections and the oral contraceptive pill have been associated with inducing flare ups.

DIAGNOSIS

There are clinical criteria combined with immunological and or biopsy criteria for making a diagnosis (e.g American College of Rheumatology (ACR) criteria, Systemic Lupus International Collaborating Clinics (SLICC) criteria).[2,3] Before the diagnosis can be confirmed, it would be prudent to exclude a vast number of differential diagnoses which may overlap with other infective and rheumatological conditions. Furthermore, other autoimmune diseases e.g. mixed connective tissue disease, diabetes type 1 and auto-immune thyroid disease can coexist with SLE.

The diagnosis of SLE for this case was made based on a positive family history of SLE, typical clinical (butterfly facial rash or Malar rash, widespread livedo reticularis rash over upper limbs and discoid lesions over the extensor surface of the right elbow extensor surface and small punctate ulcers at the roof of the soft palate) and laboratory findings (elevated ESR/CRP, low complement levels, evidence of Coomb's +ve haemolysis, worsening pancytopenia on serial blood tests, Positive immunologic markers- ANA +ve (homogeneous pattern, titer > 1:1280), Anti-DNA Ab > 200 IU, +ve Anti- SM and Anti RNP Abs, (negative anti SCL70 Ab, rheumatoid, ENA and ANCA serology), Isolated intrinsic factor abnormality in coagulation pathway-raised APTT which was not corrected on a mixing study).

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This case highlights the early onset diagnosis of SLE presenting in a young male patient who has an affected sibling who presented with similar clinical and laboratory features but had progressed to severe disease on heavy immunosuppression. The patient's condition was resistant to a combination of oral steroids and HCQ therapy but improved clinically with pulsed methylprednisolone therapy and was discharged with a weaning dose of steroids, and early Rheumatology as well as Nephrology OPD review.

It is paramount to ask about family history during the medical consultation and to be aware of the potential multisystem and heterogeneous phonotypical expression of SLE. Optimal management of this condition includes careful monitoring of these patients for disease activity, development of comorbidities and drug toxicity. Further research would be important to document the prevalence and familial clustering of SLE in Brunei in addition to the environmental and genetic factors involved in the aetiology of this condition.

An online support network can be obtained at http://www.lupusmalaysia.org/en/patients-guide/sle-a-guide-for-patients

REFERENCES

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